Crouzonodermoskeletal syndrome

Crouzonodermoskeletal syndrome is a disorder characterized by the premature joining of certain bones of the skull (craniosynostosis) during development and a skin condition called acanthosis nigricans.

The signs and symptoms of Crouzonodermoskeletal syndrome overlap with those of a similar condition called Crouzon syndrome. Common features include premature fusion of the skull bones, which affects the shape of the head and face; wide-set, bulging eyes due to shallow eye sockets; eyes that do not point in the same direction (strabismus); a small, beaked nose; and an underdeveloped upper jaw. People with Crouzon syndrome or Crouzonodermoskeletal syndrome usually have normal intelligence.

Several features distinguish Crouzonodermoskeletal syndrome from Crouzon syndrome. People with Crouzonodermoskeletal syndrome have acanthosis nigricans, a skin condition characterized by thick, dark, velvety skin in body folds and creases, including the neck and underarms. In addition, subtle changes may be seen in the bones of the spine (vertebrae) on x-rays. Noncancerous growths called cementomas may develop in the jaw during young adulthood.

Frequency

Crouzonodermoskeletal syndrome is rare; this condition is seen in about 1 person per million.

Genetic Changes

Mutations in the *FGFR3* gene cause Crouzonodermoskeletal syndrome.

The *FGFR3* gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. It remains unclear how a mutation in the *FGFR3* gene leads to the characteristic features of Crouzonodermoskeletal syndrome. This genetic change appears to disrupt the normal growth of skull bones and affect skin pigmentation.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In some cases, an affected person inherits the mutation from one affected parent. More commonly, this condition results from new mutations in the gene. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- Crouzon syndrome with acanthosis nigricans

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Crouzon syndrome with acanthosis nigricans
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2677099/

Other Diagnosis and Management Resources

- GeneReview: FGFR-Related Craniosynostosis Syndromes
  https://www.ncbi.nlm.nih.gov/books/NBK1455
- MedlinePlus Encyclopedia: Acanthosis Nigricans
  https://medlineplus.gov/ency/article/000852.htm
- MedlinePlus Encyclopedia: Craniosynostosis
  https://medlineplus.gov/ency/article/001590.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Acanthosis Nigricans
  https://medlineplus.gov/ency/article/000852.htm
- Encyclopedia: Craniosynostosis
  https://medlineplus.gov/ency/article/001590.htm
- Health Topic: Craniofacial Abnormalities
  https://medlineplus.gov/craniofacialabnormalities.html

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke
  https://www.ninds.nih.gov/Disorders/All-Disorders/Craniosynostosis-Information-Page

Educational Resources

- Center for Craniofacial Development and Disorders, Johns Hopkins Medicine
  http://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/pediatric_neurosurgery/conditions/craniosynostosis/
- Disease InfoSearch: Crouzon syndrome with acanthosis nigricans
  http://www.diseaseinfosearch.org/Crouzon+syndrome+with+acanthosis+nigricans/8115
- MalaCards: crouzon syndrome with acanthosis nigricans
  http://www.malacards.org/card/crouzon_syndrome_with_acanthosis_nigricans
- Orphanet: Crouzon syndrome-acanthosis nigricans syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=93262
- Seattle Children's Hospital and Regional Medical Center
  http://www.seattlechildrens.org/medical-conditions/chromosomal-genetic-conditions/crouzon-syndrome/
- UC Davis Children's Hospital
  https://www.ucdmc.ucdavis.edu/children/clinical_services/cleft_craniofacial/anomalies/crouzon.html

Patient Support and Advocacy Resources

- Children's Craniofacial Association
  http://www.ccakids.com
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/craniofa.html

GeneReviews

- FGFR-Related Craniosynostosis Syndromes
  https://www.ncbi.nlm.nih.gov/books/NBK1455

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Crouzonodermoskeletal+syndrome%22+OR+%22Craniofacial+Dysostosis%22+OR+%22Crouzon%27s+Disease%22+OR+%22Crouzons+Disease%22+OR+%22Craniosynostosis%22+OR+%22Crouzon+Disease%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28crouzonodermoskeletal+syndrome%29+OR+%28crouzon+syndrome+with+acanthosis+nigricans%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- CROUZON SYNDROME WITH ACANTHOSIS NIGRICANS
  http://omim.org/entry/612247

Sources for This Summary

- Cohen MM Jr. Let's call it "Crouzonodermoskeletal syndrome" so we won't be prisoners of our own conventional terminology. Am J Med Genet. 1999 May 7;84(1):74.
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  https://www.ncbi.nlm.nih.gov/books/NBK1455
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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11426459
- Vajo Z, Francomano CA, Wilkin DJ. The molecular and genetic basis of fibroblast growth factor receptor 3 disorders: the achondroplasia family of skeletal dysplasias, Muenke craniosynostosis, and Crouzon syndrome with acanthosis nigricans. Endocr Rev. 2000 Feb;21(1):23-39. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10696568

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